

(12) INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(19) World Intellectual Property  
Organization  
International Bureau



(43) International Publication Date  
21 April 2005 (21.04.2005)

PCT

(10) International Publication Number  
**WO 2005/035725 A2**

(51) International Patent Classification<sup>7</sup>:

C12N

(21) International Application Number:

PCT/US2004/033175

(22) International Filing Date: 8 October 2004 (08.10.2004)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

60/509,775 8 October 2003 (08.10.2003) US

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(81) Designated States (unless otherwise indicated, for every kind of national protection available): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NA, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.

(84) Designated States (unless otherwise indicated, for every kind of regional protection available): ARIPO (BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PL, PT, RO, SE, SI, SK, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

Published:

— without international search report and to be republished upon receipt of that report

For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.

WO 2005/035725 A2

(54) Title: METHODS FOR PRENATAL DIAGNOSIS OF CHROMOSOMAL ABNORMALITIES

(57) Abstract: Chromosomal abnormalities are responsible for a significant number of birth defects, including mental retardation. The present invention is related to methods for non-invasive and rapid, prenatal diagnosis of chromosomal abnormalities based on analysis of a maternal blood sample. The invention exploits the differences in DNA between the mother and fetus, for instance differences in their methylation states, as a means to enrich for fetal DNA in maternal plasma sample. The methods described herein can be used to detect chromosomal DNA deletions and duplications. In a preferred embodiment, the methods are used to diagnose chromosomal aneuploidy and related disorders, such as Down's and Turner's Syndrome.